METHODS FOR DIAGNOSIS AND TREATMENT OF BLOOM'S SYNDROME

Abstract of the Disclosure

The present invention provides a method for diagnosing BS as well as determining whether a subject is a carrier of a mutated *BLM* gene. The present invention also provides one or more single-stranded nucleic acid probes and antibodies which may be formulated in kits, and used for diagnosing BS or determining whether a subject is a carrier of a mutated *BLM* gene. In addition, the present invention provides a method for treating or preventing the onset of BS in a subject in need of such treatment or prevention, as well as vectors and stem cells useful for such treatment or prevention. The present invention also provides a purified and isolated nucleic acid encoding an enzymatically active BLM protein, a vector comprising this nucleic acid, a cell stably transformed with this vector, as well as a method for producing recombinant, enzymatically active BLM protein. A purified, enzymatically active BLM protein is also provided by the present invention. Finally, the present invention provides a vector, an embryonic stem cell, and a non-human, transgenic animal, each of which comprises a mutated *BLM* gene, as well as a method for producing the non-human, transgenic animal.